



NOP56 gene

NOP56 ribonucleoprotein

Normal Function

The *NOP56* gene provides instructions for making a protein called nucleolar protein 56, which is found in the nucleus of nerve cells (neurons). This protein is mostly found in neurons within an area of the brain called the cerebellum, which is involved in coordinating movements. Nucleolar protein 56 is one part (subunit) of the ribonucleoprotein complex, which is composed of proteins and molecules of RNA, DNA's chemical cousin. The ribonucleoprotein complex is needed to make cellular structures called ribosomes, which process the cell's genetic instructions to create proteins.

Located within the *NOP56* gene, in an area known as intron 1, is a string of six DNA building blocks (nucleotides); this string, known as a hexanucleotide, is represented by the letters GGCCTG and is typically repeated 3 to 14 times within intron 1. The function of this repeated hexanucleotide is unclear.

Health Conditions Related to Genetic Changes

spinocerebellar ataxia type 36

NOP56 gene mutations cause spinocerebellar ataxia type 36 (SCA36), which is a condition characterized by progressive movement problems that typically begin in mid-adulthood. In people with SCA36, the GGCCTG string in intron 1 is repeated at least 650 times.

To make proteins from the genetic instructions carried in genes, a molecule called messenger RNA (mRNA) is formed. This molecule acts as a genetic blueprint for protein production. However, a large increase in the number of GGCCTG repeats in the *NOP56* gene disrupts the normal structure of *NOP56* mRNA. Abnormal *NOP56* mRNA molecules form clumps called RNA foci within the nucleus of neurons. Other proteins become trapped in the RNA foci, where they cannot function. These proteins may be important for controlling gene activity or protein production.

Additionally, researchers believe that the large expansion of the hexanucleotide repeat in the *NOP56* gene may reduce the activity of a nearby gene called *MIR1292*. The *MIR1292* gene provides instructions for making a type of RNA that regulates the activity (expression) of genes that produce proteins called glutamate receptors. These proteins are found on the surface of neurons and allow these cells to communicate with one another. A decrease in the production of *Mir1292* RNA can lead to an increase in the production of glutamate receptors. The increased receptor

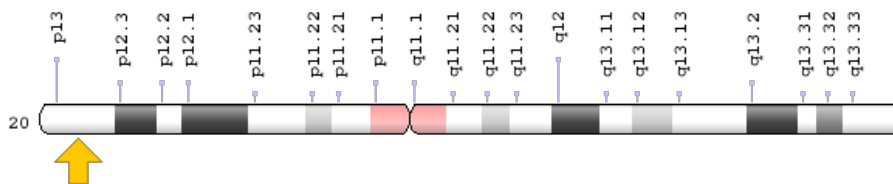
activity may overexcite neurons, which disrupts normal communication between cells and can contribute to movement difficulties.

The combination of RNA foci and overly excited neurons likely leads to the death of neurons over time. Because the *NOP56* gene is especially active in neurons in the cerebellum, these cells are particularly affected by expansion of the gene, leading to cell death in the cerebellum. Deterioration in this part of the brain leads to ataxia and the other signs and symptoms of SCA36.

Chromosomal Location

Cytogenetic Location: 20p13, which is the short (p) arm of chromosome 20 at position 13

Molecular Location: base pairs 2,652,532 to 2,658,393 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NOL5A
- NOP56 ribonucleoprotein homolog
- nucleolar protein 5A (56kDa with KKE/D repeat)
- nucleolar protein 56

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Molecular Mechanisms of TRS Instability
<https://www.ncbi.nlm.nih.gov/books/NBK6560/>

GeneReviews

- Spinocerebellar Ataxia Type 36
<https://www.ncbi.nlm.nih.gov/books/NBK231880>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NOP56%5BTIAB%5D%29+OR+%28NOL5A%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

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Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NOP56%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15911
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10528>
- UniProt
<http://www.uniprot.org/uniprot/O00567>

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Reviewed: December 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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